Impact of a Pediatric Down Syndrome Clinic on the Identification of Celiac Disease in the Patient Population

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Background

- Evidence-based outcomes of a pediatric Down syndrome (DS) clinic improving medical care access and treatment through routine screening of common comorbidities such as celiac disease are not well documented.
- There is a lack of awareness of atypical celiac disease presentations in individuals with DS.
- Routine universal screening of individuals with DS could prevent the missed or misdiagnoses.
- Within our pediatric DS clinic, celiac screening is completed every 3 years starting at the age of 3 with tTg or EMA IgA labs.

Objectives

To evaluate the impact of the pediatric DS clinic on the identification of children with DS and celiac disease using routine screening in a large patient population.

Methods

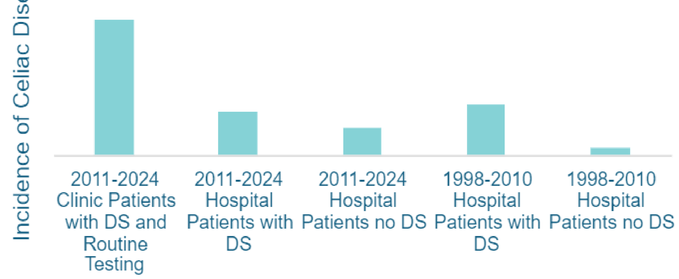
- Retrospective review of a large cohort of children with DS ages 3-22 years total=4,352 receiving care at a large pediatric hospital
  - 2011-2024 DS clinic patients=2,154
  - 2011-2024 hospital patients with DS=924
  - 1998-2010 hospital patients with DS=1,070
- Evaluated celiac data from a clinic database and electronic medical records.
- Symptoms present, type and frequency of testing completed, and other autoimmune comorbidities were reviewed.

Results

Incidence Rates

- Routine screening significantly increased the percentage of children diagnosed with celiac disease (7.4% vs. 2.4% and 2.8%) (Graph 1).

Graph 1. Incidence of celiac disease in children with and without Down syndrome

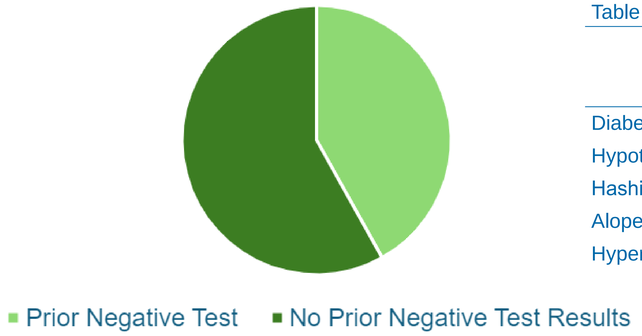


- Age at celiac diagnosis was lowest in patients with routine screening:
  - Clinic routine screening (mean = 8.6 years)
  - Clinic symptomatic testing (mean = 9.6 years)
  - 1998-2010 DS hospital patients (mean = 10.2 years)
  - 2011-2024 hospital patients (mean = 11.5 years).

Prior Testing

- For patients diagnosed with celiac disease due to routine screening with multiple documented tTg or EMA IgA results completed at CHCO, 41.9% had a prior negative result.

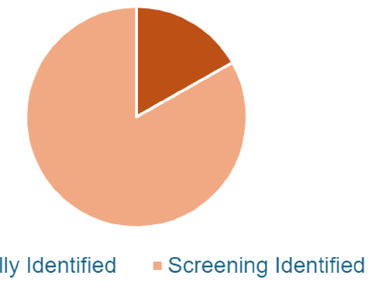
Graph 2. Prior testing history in patients diagnosed by routine screening



Symptomatic Testing

- Most patients were diagnosed with celiac disease using routine screening (83.2%) compared to identified clinically (16.8%).

Graph 3. Percentage of SCDS patients with documented diagnosis type



- The most common clinical symptoms children with DS presented with were constipation (32.6%), diarrhea (18.9%), abdominal pain (14.7%), and weight loss/failure to thrive (6.3%).
- Overall, 31.6% of patients with celiac disease were asymptomatic.

Celiac Risk Ratios

- Relative risk of celiac disease and commodities in children with DS were calculated (Table 4).
- Both hypothyroidism and diabetes were statistically significant with the largest relative risk.
- Hashimoto's had the highest relative risk, but due to small sample size, was clinically significant.

Table 4: Relative Risk of comorbidity and celiac disease

	Comorbidity and No Celiac Disease (Total=2,004) n(%)	Comorbidity and Celiac Disease (Total=160) n(%)	p-value	Relative Risk
Diabetes	12 (0.6)	4 (2.5)	p = 0.026	4.2
Hypothyroidism	574 (28.6)	68 (42.5)	p < 0.001	1.5
Hashimoto's	4 (0.2)	2 (1.3)	p = 0.067	6.3
Alopecia	32 (1.6)	5 (3.1)	p = 0.151	2
Hyperthyroidism	41 (2.0)	4 (2.5)	p = 0.571	1.2

Conclusions

- A pediatric DS clinic can identify and diagnose celiac disease sooner with routine screening implemented.
- Diagnosis of celiac disease in children with DS cannot rely on symptoms alone or traditional screening.
- Children with DS and higher risk comorbidities should be routinely tested more frequently.
- Research on asymptomatic presentations of celiac disease are not currently well studied, but new research indicates that long-term negative impacts and risk are still present.

Implications

- The pediatric DS clinic model using routine screening improves patient identification for celiac disease.
- Our results suggests that updated celiac disease recommendations in the AAP DS Guidelines should be established for this unique population.

References

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Disclosures

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